GYS2 gene

glycogen synthase 2

Normal Function

The *GYS2* gene provides instructions for making an enzyme called liver glycogen synthase. Liver glycogen synthase is produced solely in liver cells, where it helps form the complex sugar glycogen by linking together molecules of the simple sugar glucose. Glucose that is taken in from food is stored in the body as glycogen, which is a major source of energy. Glycogen that is stored in the liver can be broken down rapidly when glucose is needed to maintain normal blood sugar levels between meals.

Health Conditions Related to Genetic Changes

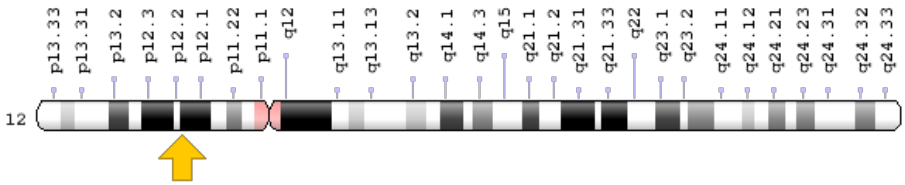
glycogen storage disease type 0

Approximately 20 mutations in the *GYS2* gene have been found to cause a form of glycogen storage disease type 0 (GSD 0) that affects the liver. Most *GYS2* gene mutations that cause this condition lead to a lack of functional glycogen synthase, resulting in a complete absence of glycogen in liver cells. Normally, glycogen is formed from the leftover glucose that is not immediately used by cells after glucose is consumed during meals. In people with GSD 0, who cannot form glycogen, the extra glucose is released by the body. As a result, people with this condition have no stored energy during long periods without food (fasting). During these periods, affected individuals may develop low blood sugar (hypoglycemia) and nausea as well as other signs and symptoms of GSD 0.

Chromosomal Location

Cytogenetic Location: 12p12.1, which is the short (p) arm of chromosome 12 at position 12.1

Molecular Location: base pairs 21,536,189 to 21,604,858 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- glycogen [starch] synthase, liver
- glycogen synthase 2 (liver)
- GYS2_HUMAN
- liver glycogen synthase
- liver glycogen synthase 2

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Glycogen Metabolism in the Liver Regulates the Blood-Glucose Level
  https://www.ncbi.nlm.nih.gov/books/NBK22444/#A2961
- Biochemistry (fifth edition, 2002): Glycogen Synthase Catalyzes the Transfer of Glucose from UDP-Glucose to a Growing Chain
  https://www.ncbi.nlm.nih.gov/books/NBK22413/#A2948

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GYS2%5BTIAB%5D%29+OR+%28glycogen+synthase+2%5BTIAB%5D%29%29+OR+%28%28liver+glycogen+synthase%5BTIAB%5D%29+OR+%28glycogen+synthase+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- GLYCOGEN SYNTHASE 2
  http://omim.org/entry/138571

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GYS2%5Bgene%5D
- HGNC Gene Family: Glycosyl transferases group 1 domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/427
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4707
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2998
- UniProt
  http://www.uniprot.org/uniprot/P54840

Sources for This Summary

- Bachrach BE, Weinstein DA, Orho-Melander M, Burgess A, Wolfsdorf JI. Glycogen synthase deficiency (glycogen storage disease type 0) presenting with hyperglycemia and glucosuria: report of three new mutations. J Pediatr. 2002 Jun;140(6):781-3.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12072888
- OMIM: GLYCOGEN SYNTHASE 2
  http://omim.org/entry/138571
- Orho M, Bosshard NU, Buist NR, Gitzelmann R, Aynsley-Green A, Blümel P, Gannon MC, Nuttall FQ, Groop LC. Mutations in the liver glycogen synthase gene in children with hypoglycemia due to glycogen storage disease type 0. J Clin Invest. 1998 Aug 1;102(3):507-15.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9691087
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC508911/

- Soggia AP, Correa-Giannella ML, Fortes MA, Luna AM, Pereira MA. A novel mutation in the glycogen synthase 2 gene in a child with glycogen storage disease type 0. BMC Med Genet. 2010 Jan 5;11:3. doi: 10.1186/1471-2350-11-3.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20051115
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2837020/
- Weinstein DA, Correia CE, Saunders AC, Wolfsdorf JI. Hepatic glycogen synthase deficiency: an infrequently recognized cause of ketotic hypoglycemia. Mol Genet Metab. 2006 Apr;87(4):284-8. Epub 2005 Dec 6. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16337419
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1474809/

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